Hirschsprung's disease associated with Mowat-Wilson syndrome

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DESCRIPTION

A term male infant was born after an uneventful pregnancy and normal vaginal delivery with a birth weight of 3480 g. There was no consanguinity among parents. He required no resuscitation at birth. He had typical dysmorphic facial features such as square-shaped face, a prominent but narrow triangular chin, deep set but large eyes, hypertelorism, saddle nose, broad nasal bridge, open mouth, everted lower lip, posteriorly rotated ears and large uplifted ear lobes with a central depression. Recognition of the characteristic facies led to referral for genetic counselling. He had delayed passage of meconium with abdominal distention and bilious vomiting on second day of life. He responded well to saline rectal washouts and rectal suction biopsy confirmed Hirschsprung's disease (figure 1). He was sent home with daily saline rectal washouts by parents until 3 months of age at which time he underwent primary Duhamel pull through operation using small hockey stick smiling left lower abdominal incision. His postoperative period was

uneventful. Genetic team on investigation found it to be sporadic resulting from de novo deletion of the ZFHX1B gene with cytogenetic deletion of 2q22-23. There was no indication of any deletion in the parental chromosomes on this identical location and therefore it was concluded that it was a de novo mutation rather than inherited pattern of the mendelian type. In addition he has glandular hypospadias, absent corpus callosum, abnormal EEG, moderate mental retardation and microcephaly. At 10-year follow-up he is progressing well, has normal bowel function and shows typical facial changes. Mowat-Wilson syndrome with Hirschsprung's disease is a complex syndromic genetic disease with multiple congenital anomalies and typical dysmorphic features with mental retardation.^{1 2} Generally, the outcome of syndromic Hirschsprung's disease in such cases is very poor.³ Our case is exceptional and a strong reminder of the fact that we should at least give definitive surgery a chance to see how they do, and meticulously performed surgery does help.

Figure 1 (A–C) Histology: (A) normal control rectal biopsy showing several clusters of ganglion cells in the submucosa (arrows); (B) Patient's rectal biopsy showing no ganglion cells in the submucosa; (C) several large nerve trunks seen in the submucosa (arrows). (D and E) Acetyl cholinesterase histochemistry: (D) normal control Ache stain. There is only a light brown blush in the muscularis mucosa. There are no large nerves (these would stain black); (E) Ache stain showing the large nerves in (C) (arrows). (F) Ache stain showing abnormal nerves in the muscularis mucosa and lamina propria (arrows).

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Learning points

- ▶ Characteristic dysmorphic features in association with Hirschsprung's disease and multiple congenital anomalies and mental retardation should alert the possibility of Mowat-Wilson syndrome.
- ▶ Genetic counselling should be conducted and other conditions should be excluded. Normal parental genes and chromosomes may suggest a de novo mutation in the offspring.
- ▶ Generally speaking the outcome of pull-through operation for Hirschsprung's disease is very poor and our case is an exception to this rule perhaps due to de novo mutation and meticulously performed single stage neonatal primary pull-through surgery.

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